



## WASHC5 gene

WASH complex subunit 5

### Normal Function

The *WASHC5* gene provides instructions for making a protein called strumpellin. Strumpellin is active (expressed) throughout the body, although its exact function is unknown. The protein's structure suggests that strumpellin may interact with the structural framework inside cells (the cytoskeleton) and may attach (bind) to other proteins.

### Health Conditions Related to Genetic Changes

#### spastic paraplegia type 8

At least three mutations in the *WASHC5* gene have been found to cause spastic paraplegia type 8. These mutations change single building blocks (amino acids) in the strumpellin protein. One mutation that has been seen in multiple families replaces the amino acid valine with the amino acid phenylalanine at position 626 in strumpellin (written Val626Phe or V626F). *WASHC5* gene mutations are thought to change the structure of the strumpellin protein. It is unknown how the altered strumpellin protein causes muscle weakness, muscle stiffness, and other features of spastic paraplegia type 8.

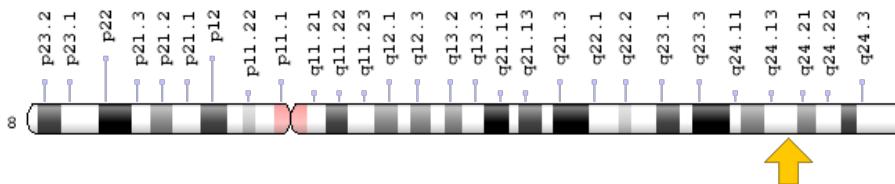
#### cancers

Research has shown that the *WASHC5* gene is abnormally active (overexpressed) in certain types of prostate cancer. Scientists do not know what causes this abnormal expression and have not determined whether the *WASHC5* gene plays a role in the development of prostate cancer.

## Chromosomal Location

Cytogenetic Location: 8q24.13, which is the long (q) arm of chromosome 8 at position 24.13

Molecular Location: base pairs 125,024,260 to 125,091,819 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- KIAA0196
- MGC111053
- SPG8
- STRUM\_HUMAN
- strumpellin

## Additional Information & Resources

### GeneReviews

- Spastic Paraplegia 8  
<https://www.ncbi.nlm.nih.gov/books/NBK1827>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28KIAA0196%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- KIAA0196 GENE  
<http://omim.org/entry/610657>

## Research Resources

- **Atlas of Genetics and Cytogenetics in Oncology and Haematology**  
[http://atlasgeneticsoncology.org/Genes/GC\\_WASHC5.html](http://atlasgeneticsoncology.org/Genes/GC_WASHC5.html)
- **ClinVar**  
<https://www.ncbi.nlm.nih.gov/clinvar?term=WASHC5%5Bgene%5D>
- **HGNC Gene Family: WASH complex**  
<http://www.genenames.org/cgi-bin/genefamilies/set/1331>
- **HGNC Gene Symbol Report**  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=28984](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=28984)
- **NCBI Gene**  
<https://www.ncbi.nlm.nih.gov/gene/9897>
- **UniProt**  
<http://www.uniprot.org/uniprot/Q12768>

## **Sources for This Summary**

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*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16130124>

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Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/gene/WASHC5>

Reviewed: March 2009  
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
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